



potassium-aggravated myotonia

Potassium-aggravated myotonia is a disorder that affects muscles used for movement (skeletal muscles). Beginning in childhood or adolescence, people with this condition experience bouts of sustained muscle tensing (myotonia) that prevent muscles from relaxing normally. Myotonia causes muscle stiffness that worsens after exercise and may be aggravated by eating potassium-rich foods such as bananas and potatoes. Stiffness occurs in skeletal muscles throughout the body. Potassium-aggravated myotonia ranges in severity from mild episodes of muscle stiffness to severe, disabling disease with frequent attacks. Unlike some other forms of myotonia, potassium-aggravated myotonia is not associated with episodes of muscle weakness.

Frequency

This condition appears to be rare; it has been reported in only a few individuals and families worldwide.

Genetic Changes

Mutations in the *SCN4A* gene cause potassium-aggravated myotonia.

The *SCN4A* gene provides instructions for making a protein that is critical for the normal function of skeletal muscle cells. For the body to move normally, skeletal muscles must tense (contract) and relax in a coordinated way. Muscle contractions are triggered by the flow of positively charged atoms (ions), including sodium, into skeletal muscle cells. The *SCN4A* protein forms channels that control the flow of sodium ions into these cells.

Mutations in the *SCN4A* gene alter the usual structure and function of sodium channels. The altered channels cannot properly regulate ion flow, increasing the movement of sodium ions into skeletal muscle cells. The influx of extra sodium ions triggers prolonged muscle contractions, which are the hallmark of myotonia.

Inheritance Pattern

Potassium-aggravated myotonia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In some cases, an affected person inherits a mutation in the *SCN4A* gene from one affected parent. Other cases result from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- PAM
- sodium channel myotonia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Potassium aggravated myotonia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0752355/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Drugs and Supplements: Acetazolamide
<https://medlineplus.gov/druginfo/meds/a682756.html>
- Health Topic: Muscle Disorders
<https://medlineplus.gov/muscledisorders.html>

Genetic and Rare Diseases Information Center

- Potassium aggravated myotonia
<https://rarediseases.info.nih.gov/diseases/4459/potassium-aggravated-myotonia>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
<https://www.ninds.nih.gov/Disorders/All-Disorders/Myotonia-Information-Page>

Educational Resources

- Disease InfoSearch: Potassium aggravated myotonia
<http://www.diseaseinfosearch.org/Potassium+aggravated+myotonia/5901>
- Orphanet: Potassium-aggravated myotonia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=612

Patient Support and Advocacy Resources

- Muscular Dystrophy Association
<https://www.mda.org/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/muscular.html>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22potassium-aggravated+myotonia%22+OR+%22Myotonic+Disorders%22+OR+%22Myotonia+Fluctuans%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28potassium-aggravated+myotonia%5BTIAB%5D%29+OR+%28myotonia+fluctuans%5BTIAB%5D%29+OR+%28myotonia+permanens%5BTIAB%5D%29%29+OR+%28%28acetazolamide%5BTIAB%5D%29+AND+%28myotonia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MYOTONIA, POTASSIUM-AGGRAVATED
<http://omim.org/entry/608390>

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